

# Case Report of Mayer-Rokitansky-Kuster-Hauser Syndrome associated with Renal Anomaly Diagnosed Incidentally in a Patient with Acute Abdomen

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**Abstract:** *Mayer-Rokitansky-Kuster-Hauser syndrome, also known as Rokitansky syndrome, is a malformation of the female genital tract that is due to interrupted embryonic development of the paramesonephric (müllerian) ducts. It leads to hypoplasia of the uterus and the upper two-thirds of the vagina. It affects approximately one in 4500 live births (1). It occurs as a purely genital malformation (type 1), but also with associated malformations (type 2 and MURCS association; Mullerian Renal, Cervicothoracic Somite abnormalities.<sup>2</sup> Malformations of the kidneys and urinary tract the skeleton, and more rarely of the heart and central nervous system have been described.<sup>3</sup>We are reporting a case of incidental finding of MRKH syndrome with associated horseshoe kidney in a 29-year-old female, who came to our emergency department with sudden onset of pain right iliac fossa which is progressive and is non radiating. The clinical diagnosis of Acute appendicitis was kept with MANTERAL score of 5. CECT Abdomen was done in emergency with finding of medially oriented inferior pole of bilateral kidney which is fused in midline at L3 vertebral body level with non visualised uterus and cervix. Appendix is normal. Thus diagnosis of MRKH syndrome is kept with unusual clinical presentation.*

**Keywords:** paramesonephric, mullerian, ducts, Mullerian Renal, Cervicothoracic Somite

## 1. Introduction

Mayer-Rokitansky-Kuster-Hauser syndrome is a malformation of the female genital tract that is due to interrupted embryonic development of the paramesonephric (Mullerian) ducts. It leads to hypoplasia of the uterus and the upper two-thirds of the vagina. It affects approximately one in 4500 live births.<sup>1</sup> Ovarian function is normal; so patient present during adolescence with primary amenorrhea in the presence of normal pubertal development and secondary sexual characteristics.<sup>1</sup> It occurs as a purely genital malformation (type 1), also associated malformation (type 2) and MURCS (Mullerian renal, cervicothoracic somite abnormalities).<sup>2</sup> Malformations of the kidneys and urinary tract, skeleton and rarely of the heart and central nervous system have been described.<sup>3</sup>

We are reporting a case of incidental finding of MRKH syndrome with associated horseshoe kidney in a 29-year-old female, who came to our emergency department with sudden onset of pain right iliac fossa which is progressive and is non

radiating. The clinical diagnosis of Acute appendicitis was kept with MANTERAL score of 5. CECT Abdomen was done in emergency with finding of medially oriented inferior pole of bilateral kidney which is fused in midline at L3 vertebral body level with non visualised uterus and cervix. Appendix is normal.

## 2. Case Report

A 29-year-old female, who came to our emergency department with sudden onset of pain in right iliac fossa which is progressive in nature and is non radiating. The clinician kept the diagnosis of Acute appendicitis was with MANTERAL score of 5. Ultrasound was done in emergency by resident and normal report was given. To rule out acute appendicitis CECT Abdomen was done in emergency. CECT abdomen finding was medially oriented inferior pole of bilateral kidney which is fused in midline at L3 vertebral body level with non visualised uterus and cervix. Appendix is normal. Thus final diagnosis of MRKH syndrome is kept.



**Figure 1:** CECT Abdomen axil image show fused inferior pole of bilateral kidney in midline ( Horseshue Kidney).



**Figure 2 (a)**



**Figure 2 (b)**

Figure 2(a-b): CECT Abdomen axilimage( Fig.2a) and sagittal ( Fig 2b) shows absent uterus and cervix.

### 3. Discussion

The incidence of MRKH in ~1:4,500 live female births, congenital renal malformations (as well as unilateral renal agenesis) in MRKH patients are higher compared to the general population. This is not surprising due to the association and interaction of the two ductal systems for normal genital and renal development. Combined urogenital malformations are common with estimations of 10 in 100 cases and account for over 30% of all congenital malformations.<sup>4</sup> Malformations of the genital and renal axis are common, e.g. 35% of females where unilateral renal agenesis showed partial or complete duplication of the genital tract.<sup>5</sup> renal agenesis was present in 43% of patients with uterus didelphys and 10% of patients with other genital tract abnormalities had an abnormal or ectopic kidney.<sup>6</sup> Asour case is incidentally diagnosed in a patient of acute pain abdomen with absent uterocervix and horseshoe kidney so in all patients with genital malformations should be evaluated for renal abnormalities, as well as patient with renal abnormalities should be assessed for genital malformations.

### 4. Conclusion

In conclusion, patient with absent uterocervix, müllerian agenesis, renal abnormalities should also be assessed as MRKH syndrome also associated with renal abnormality like horseshoe kidney as our patient. Though MRI is best modality to evaluate MRKH syndrome patient, our case was diagnosed in emergency with complain of acute abdomen.

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